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Leukaemia Section
Short Communication

dic(9;18)(p13;q11) PAX5/ZNF521

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Identity

Note
See also the paper on dic(9;20)(p11-13;q11).

Clinics and pathology

Disease
Acute lymphoblastic leukaemia (ALL).

Phenotype/cell stem origin
B-cell precursor ALL.

Epidemiology
One case to date (Mullighan et al., 2007).

Prognosis
No data.

Genes involved and proteins

PAX5
Location
9p13.2

Protein
Lineage-specific transcription factor; recognizes the concensus recognition sequence GNCCANTGAAGCGTGAC, where N is any nucleotide. Involved in B-cell differentiation. Entry of common lymphoid progenitors into the B cell lineage depends on E2A, EBF1, and PAX5; activates B-cell specific genes and repress genes involved in other lineage commitments. Activates the surface cell receptor CD19 and repress FLT3. Pax5 physically interacts with the RAG1/RAG2 complex, and removes the inhibitory signal of the lysine-9-methylated histone H3, and induces V-to-DJ rearrangements. Genes repressed by PAX5 expression in early B cells are restored in their function in mature B cells and plasma cells, and PAX5 repressed (Fuxa et al., 2004; Johnson et al., 2004; Zhang et al., 2006; Cobaleda et al., 2007).

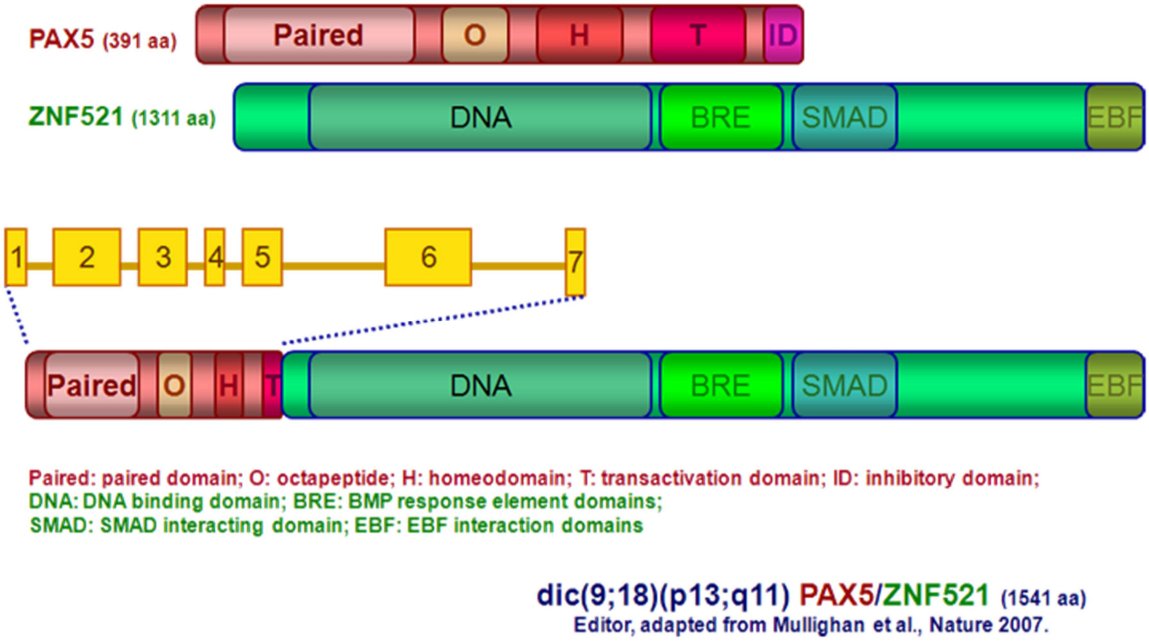
ZNF521
Location
18q11

Protein
Transcription factor. Involved in B-cell lineage commitment, in the differentiation of neural progenitors; Inhibits EBF1. Binds Runx2 and represses its transcriptional activity. Regulates osteoblast differentiation and bone formation (Bond et al., 2008; Lobo et al., 2008; Wu et al., 2009; Hesse et al., 2010).

Result of the chromosomal anomaly

Hybrid gene
Description
Fusion of PAX5 exon 7 to ZNF521 exon 4.

Fusion protein



Description

The predicted fusion protein contains the DNA binding paired domain of PAX5 and the DNA-binding and transcriptional regulator domain of ZNF521. 1541 amino acids.

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